SLC19A2 gene

solute carrier family 19 member 2

Normal Function

The *SLC19A2* gene provides instructions for making a protein called thiamine transporter 1. This protein is located on the surface of cells, where it works to bring vitamin B1 (thiamine) into cells. Thiamine helps the body convert carbohydrates into energy, and it is also essential for the functioning of the heart, muscles, and nervous system. This vitamin must be obtained from the diet because the body cannot produce thiamine on its own. Many different foods contain thiamine, including whole grains, pasta, fortified breads and cereals, lean meats, fish, and beans.

Health Conditions Related to Genetic Changes

thiamine-responsive megaloblastic anemia syndrome

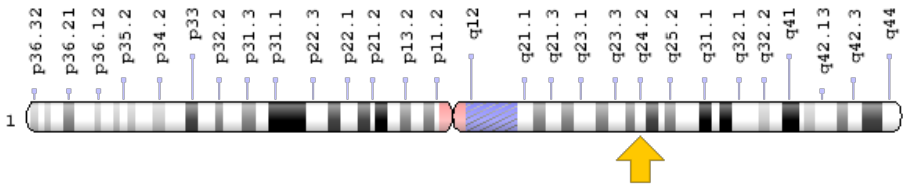
At least 17 mutations in the *SLC19A2* gene have been found to cause thiamine-responsive megaloblastic anemia syndrome. Most of these mutations lead to the production of an abnormally short, nonfunctional thiamine transporter 1. Other mutations change single protein building blocks (amino acids) in thiamine transporter 1, which disrupts the proper folding of the protein or prevents it from reaching the cell surface. All of these mutations prevent thiamine transporter 1 from bringing thiamine into the cell.

It remains unclear how the absence of thiamine transporter 1 leads to the seemingly unrelated symptoms of megaloblastic anemia, diabetes, and hearing loss. Research suggests that an alternative method for transporting thiamine is present in all the cells of the body, except where blood cells and insulin are formed (in the bone marrow and pancreas, respectively) and cells in the inner ear.

Chromosomal Location

Cytogenetic Location: 1q24.2, which is the long (q) arm of chromosome 1 at position 24.2

Molecular Location: base pairs 169,463,909 to 169,485,970 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- high affinity thiamine transporter
- reduced folate carrier protein (RFC) like
- S19A2_HUMAN
- solute carrier family 19 (thiamine transporter), member 2
- solute carrier family 19, member 2
- TC1
- thiamine transporter 1
- THT1
- THTR1
- TRMA

Additional Information & Resources

GeneReviews

- Thiamine-Responsive Megaloblastic Anemia Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1282

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC19A2%5BTIAB%5D%29+OR+%28%28high+affinity+thiamine+transporter%5BTIAB%5D%29+OR+%28thiamine+transporter+1%5BTIAB%5D%29+OR+%28THT1%5BTIAB%5D%29+OR+%28THTR1%5BTIAB%5D%29+OR+%28TRMA%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 2
  http://omim.org/entry/603941

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC19A2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC19A2%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10938
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/10560
- UniProt
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